This week in the BMJ

Genes and environment determine survival in familial hypercholesterolaemia

The risk of early death and premature cardiovascular disease in familial hypercholesterolaemia may have been overestimated because previous studies have looked only at patients and families who sought medical attention. Sijbrands et al (p 1019) traced all members of a Dutch pedigree dating from 1800 to 1989 and calculated their risk of death. They found that many untreated patients had normal life spans. The variation in mortality, they conclude, suggests that outcome is determined by complex interaction between genetic and environmental factors.

Predictive genetic tests can save lives but utility varies because most have uncertain predictive value

Predictive genetic tests are used to identify individuals at risk of developing a particular disease. Such tests will be increasingly available as the genetic factors that underpin common diseases are identified. On p 1052 Evans et al explore the utility of such testing. They say that, although predictive genetic testing has the potential to save lives through targeted surveillance and preventive measures, application can be complex and even potentially harmful to patients. More discussion is needed about testing which needs to be tailored to individual needs and preferences.

Genetic medicine needs to be integrated into primary care

Advances in genetic medicine and the predicted rise in demand for genetic advice and information mean that primary care practitioners will need to become genetically literate. On p 1027 Emery and Hayflick discuss how genetic medicine is beginning to permeate routine practice. Incorporating genetics into primary care will require innovative educational initiatives. The earliest impact of genetic medicine in primary care is likely to be in pharmacogenetics, the science of identifying those who will respond to drugs and those who are likely to get side effects. This should make drug therapy safer and more effective.

This card is fiction but some think that predictive testing for common diseases may not be far off

Knowledge of genetic risk may not change behaviour

Genetic developments make it increasingly likely that people will be given DNA based information about their chances of developing common preventable diseases such as cancer and heart disease. Marteau and Lerman (p 1056) examine the assumption that telling people about genetic risk will motivate them to change their behaviour. From the limited evidence available they conclude that people are not likely to change their behaviour in response to genetic risk information and in some cases information may have the undesirable effect of reducing the motivation to change.

Regional genetic centres could lead new service developments

Specialist geneticists in the UK have built up considerable expertise in managing patients at high risk of genetic conditions through a collaborative network of regional genetic centres. On p 1048 Donnai and Elles describe the services these centres provide and suggest that they are well placed to adapt to coordinate the provision of new genetic services and educational initiatives in all branches of medicine. The introduction of well trained genetic counsellors is seen as crucial to the successful integration of services into primary care.

Genotyping may become routine in clinical practice

The way in which new genetic technologies are being used to search for genes that determine susceptibility to common disease, and the rationale for doing this, is described by Mathew (p 1031). Looking to the future, he illustrates how practice will change with a hypothetical case of a 45 year old woman presenting with hypertension. A buccal smear is taken and her genotype identified before she is prescribed a specific drug and dietary regimen known to be effective in her particular subtype of hypertension.